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Case report

Cleidocranial dysplasia with bilateral posterior glenohumeral dislocation: A case-report



A. Hardy^{a,*}, M.W. Thiong'o^a, A. Leroy^a, P. Hardy^b, C. Laporte^a

^a Service de chirurgie orthopédique, centre hospitalier de Meaux, 6-8, rue Saint-Fiacre, 77100 Meaux, France

^b Service de chirurgie orthopédique, hôpital Ambroise-Paré, 9, avenue Charles-de-Gaule, 92100 Boulogne-Billancourt, France

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ABSTRACT

A 31-year-old man experienced bilateral posterior glenohumeral dislocation during seizures. He had cleidocranial dysplasia with complete absence of both clavicles. Cleidocranial dysplasia is a rare inherited disease also known as Marie-Sainton syndrome and responsible for dental abnormalities well-known to stomatologists and dentists. Other manifestations include defective development of the skull bones and hypoplastic or aplastic clavicles. We found no previous reports of bilateral posterior glenohumeral dislocation in patients with cleidocranial dysplasia. The objective of this work was to look for an association between clavicular aplasia and posterior glenohumeral dislocation.

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1. Introduction

We report a case of acute, traumatic, posterior dislocation of both glenohumeral joints during seizures in a patient with bilateral clavicular agenesis. The possibility that an association may exist between clavicular agenesis and bilateral posterior glenohumeral dislocation is discussed.

2. Case-report

A 31-year-old man receiving follow-up for cleidocranial dysplasia (CCD) was admitted for a staphylococcal infection of the face believed to be a complication of recent surgery to extract supernumerary teeth. He had three febrile seizures associated with alcohol withdrawal and probably related to delirium tremens. The clinical evaluation indicated posterior dislocation of both shoulders. Radiographs confirmed this diagnosis, showing posterior dislocation of both glenohumeral joints, as well as anteromedial humeral head impaction fractures (McLaughlin fractures) (Fig. 1A and B). Bilateral clavicular agenesis was a previously undiagnosed feature of the CCD. He denied any history of anterior or posterior shoulder instability. Appropriate measures were taken to ensure resolution of the seizures and delirium tremens.

Relocation of the shoulders was then achieved in the operating room, under general anaesthesia, using external manoeuvres

(traction, external rotation, and abduction). Joint testing after successful shoulder relocation showed marked instability with internal rotation greater than 90°. Computed tomography (CT) visualised anterior humeral notches measuring 3 × 2.5 cm and 4 × 2.5 cm, respectively (Fig. 2A and B). The P/R index [1,2], used by extension and defined as the depth of the humeral notch over the radius of the humeral head, was 77%. CT showed no dysplastic or other abnormalities of the glenoid cavity and confirmed the complete absence of both clavicles. Retroversion of the glenoid cavity was within the normal range at the superior and inferior poles and was slightly increased in the middle: from top to bottom, retroversion was −9°, −13°, and −15°; compared to reported normal values [3] of −5° (range, −2° to −15°), −2° (range, 0° to −8°), and −9° (range, −2° and −15°) (Fig. 3). Cerebral CT showed a persistently open anterior fontanelle.

Non-operative treatment was deemed the best option. The shoulders were immobilised in 10° of external rotation with the elbows by the side, in thoracobrachial casts (Fig. 4), then by a thoracic brace with an arm and forearm gutter on each side. Total duration of immobilisation was 6 weeks.

A family study identified a single other case of CDD, in the father. The mother and two siblings, a sister and a brother, had no evidence of the disease. Neither were any cases known in the family (Fig. 5).

At last follow-up after 1 year, the patient reported no recurrent shoulder dislocation despite having experienced another seizure. Neither did he report any pain or sensation of shoulder instability. Findings by physical examination included slight motion-range alterations, without pain or muscle strength impairments, and an abnormally small bioacromial diameter. The bilateral clavicular

* Corresponding author. Tel.: +33 615 101 016.

E-mail address: Alexandre.hardy@me.com (A. Hardy).

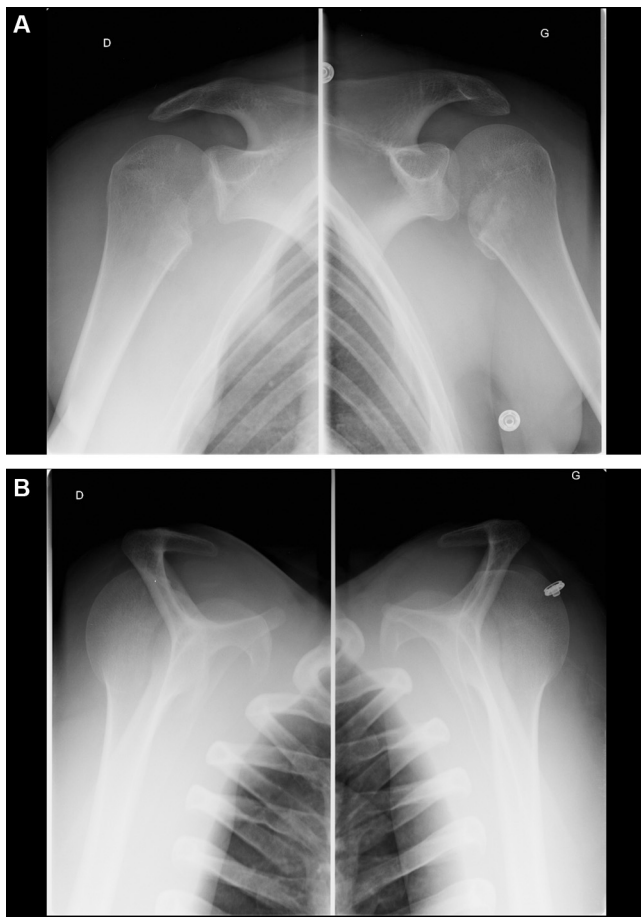


Fig. 1. Acute bilateral posterior glenohumeral dislocation at admission. A. Antero-posterior radiograph of both shoulders. B. Lateral radiographs of both shoulders.

agenesis resulted in abnormal scapular winging. The patients reported no apprehension during anterior or posterior shoulder movements.

3. Discussion

CCD is an inherited disease also known as Marie-Sainton syndrome. The cause is a mutation in the *RUNX2* gene on chromosome 6p21. *RUNX2* encodes a transcription factor required for osteoblast differentiation [4]. CCD is inherited on an autosomal dominant basis with variable penetrance. Its prevalence is 1/1,000,000. One-fourth of patients have de novo mutations with unaffected parents [4,5]. The mutation impairs the intramembranous ossification process, most notably at the skull and clavicles. In most individuals, ossification starts with the clavicles, at 5 to 6 postconceptional weeks. The spectrum of the clavicular ossification impairment includes hypoplasia, hypocalcification, and complete agenesis. The main clinical features are found at the skull and consist of prominent frontal and occipital bones, maxillary hypoplasia with dental abnormalities (particularly supernumerary teeth), brevity or absence of the nasal bones, sinus abnormalities, and an open anterior fontanelle [6]. This last feature was present in our patient (Fig. 6). Complete clavicular agenesis is seen in 10% of patients, who have sloping and hypermobile shoulders [7], a small biacromial diameter, and the ability to approximate the shoulders anteriorly [8–12]. This hypermobility does not seem to promote instability of the glenohumeral joint, since reports of shoulder dislocation as a complication of CDD are scarce and always consist of anterior dislocation [12,13].

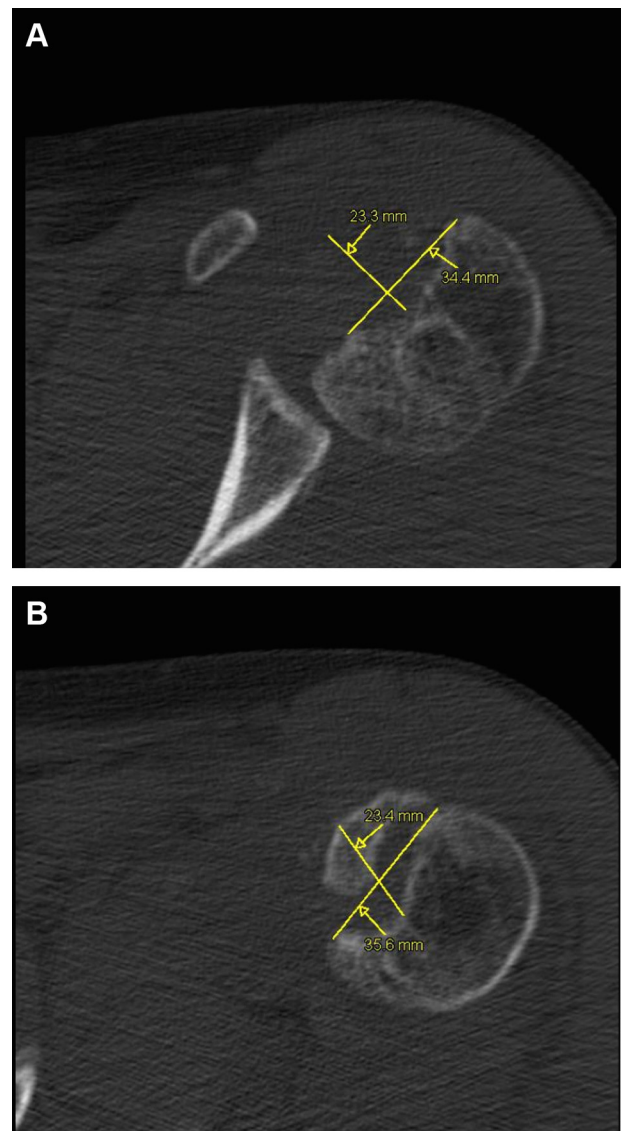


Fig. 2. Computed tomography of the shoulder. A. View centred on the notch at the level of the glenoid cavity. B. View centred on the notch at the distal end of the lesion.

The original feature of our case-report is the previously unpublished combination of posterior glenohumeral dislocation and bilateral clavicular agenesis. Thus, few orthopaedic surgeons are familiar with CCD.

Bilateral posterior glenohumeral dislocation is rare but well documented (61 case-reports identified in the international literature, with the first having been published in 1946 [14]). The main mechanisms responsible for this injury are seizures (epilepsy, tonic-clonic movements, etc.), electrocution, and high-energy trauma. Reduction under general anaesthesia is the most widely used treatment approach. Once reduction has been achieved, surgery may deserve consideration if the notch size exceeds 30% of the humeral head surface by CT. Surgery then involves lifting up the impaction and grafting to fill the defect, via the deltopectoral approach [15].

Congenital absence of the clavicle results in abnormally pronounced scapular winging, which may result in posterior glenohumeral instability, simply by altering the functional position of the glenoid cavity in space. Another likely factor is regional ligament dysplasia due to the absence of clavicular ossification. Thus, the ligaments may fail to provide adequate support, particularly when the shoulders roll into maximal internal rotation, for instance during

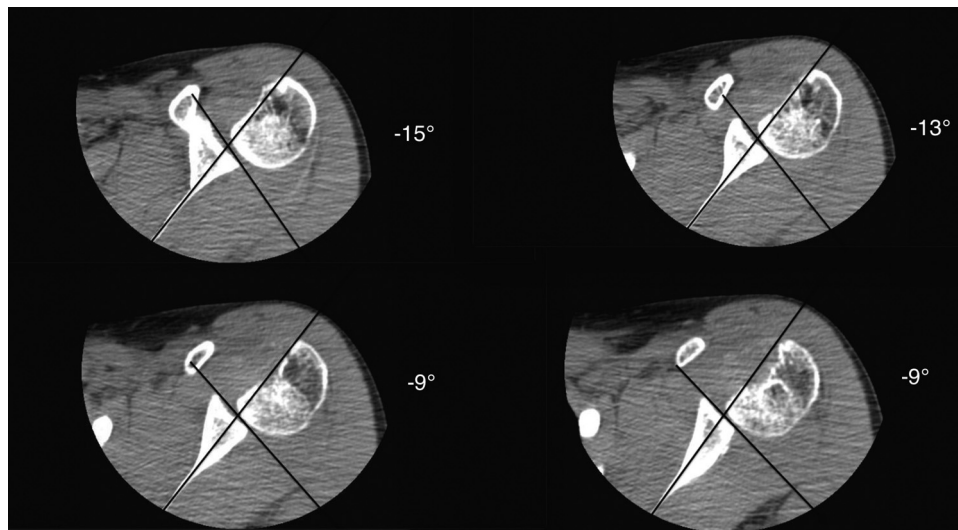


Fig. 3. Measurement of glenoid cavity retroversion on computed tomography images taken at different heights.

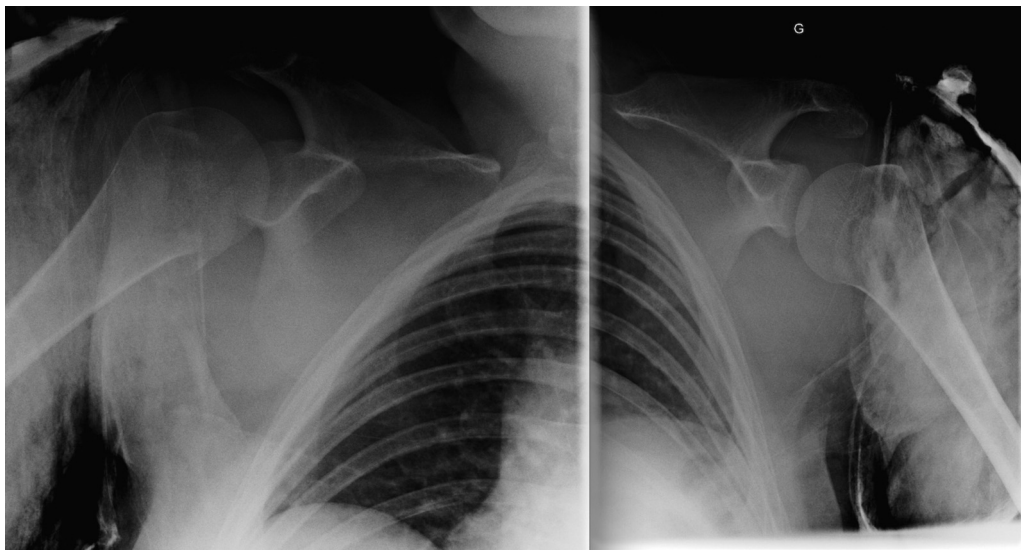


Fig. 4. Follow-up radiograph after immobilisation of the shoulders in a thoracobrachial cast.

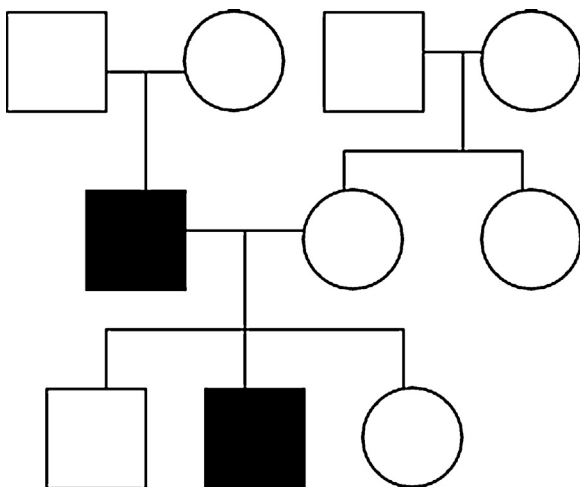


Fig. 5. Pedigree of the patient.

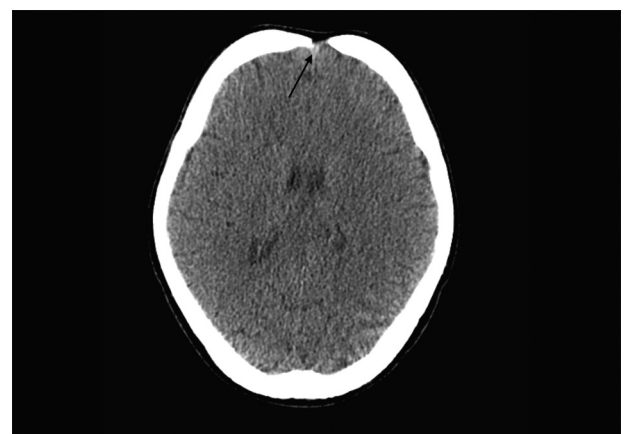


Fig. 6. Cerebral computed tomography: note the open anterior fontanelle.

seizures. In normal individuals, the clavicle may limit the range of internal shoulder rotation, thereby acting as a barrier against posterior dislocation. The small biacromial diameter combined with the narrow shoulders and small scapulae result in sloping and hypermobile shoulders. Acquired absence of the clavicle does not seem to increase the risk of posterior shoulder dislocation, as this event has not been reported after clavicle removal during chest or vascular surgery.

Thus, whether the CDD contributed to the occurrence of bilateral posterior glenohumeral dislocation in our patient remains unclear. In addition, CCD is extremely rare and case-series are therefore difficult to assemble. Nevertheless, the combination of seizures and scapular winging in a patient with clavicular agenesis and shoulder hypermobility supports a relationship between the CDD and the bilateral shoulder dislocation.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

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